CASE REPORT

Obstructive Sleep Apnoea (OSA) in a Case of Oculo-Auriculo-Vertebral Spectrum (OAVS)

Dipti Gothi, J. M. Joshi

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See end of article for authors affiliations

Address for correspondence:

Dipti Gothi, J. M. Joshi
Department of Respiratory Medicine,
T. N. Medical College and B. Y. L.
Nair Hospital, Mumbai, India.

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ABSTRACT

We report a 3 year old girl with Oculo-auriculo-vertebral spectrum also called the Goldenhar's syndrome presenting with microtia, hemifacial microsomia, cardiac anomaly and obstructive sleep apnoea. Sleep related breathing disorders in children have received considerable attention in recent times. Obstructive sleep apnoea in OAVS has been has been reported rarely and is due to aplastic mandible or micrognathia. Polysomnography confirmed obstructive sleep apnoea enabling an appropriate surgical therapy.

Keywords: Oculo-auriculo-vertebral spectrum (OAVS), Goldenhar's syndrome, Obstructive sleep apnoea (OSA)

Introduction

Oculo-auriculo-vertebral dysplasia also called the Goldenhar's syndrome or the Goldenhar-Gorlin syndrome is characterized by anomalies of the eye, ear (mostly microtia), hemifacial microsomia, and defects of the vertebral column. The criteria described earlier for the diagnosis of Goldenhar's syndrome consisted of an eye abnormality (lipoma, lipodermoid, epibulbar dermoid, or upper eyelid coloboma) associated with ear, mandibular, or vertebral anomalies (two of the three).[1] However, recently the clinical data and photographs of patients with were evaluated, all presenting with either isolated microtia or preauricular tags in association with hemifacial microsomia as minimal diagnostic criteria. Based on the main clinical findings and unilateral or bilateral involvement, a new classification system consisting of six subgroups i.e. oculo-auriculo-vertebral spectrum (OAVS) has been developed.[2] Sleep related breathing disorders in children have received considerable attention in recent times. The estimated prevalence of snoring in children is 3 to 12 percent, while OSA affects 1 to 10 percent of pediatric population.[3] Obstructive sleep apnoea (OSA) in OAVS has been has been reported rarely and is due to aplastic mandible or micrognathia.[4,5]. We report a 3 year girl of OAVS with OSA due to hypoplasia of the mandible.

Case Report

A 3-year-old girl with rudimentary right ear and facial asymmetry (Figure 1A, 1B) since birth was referred for evaluation of snoring, nocturnal choking, frequent arousals, nocturia and witnessed apnoea of two and a half year duration. She did not have excessive daytime sleepiness. She was born by cesarean section without any birth asphyxia, birth trauma, or neonatal insult. There was no family history of similar abnormality. Examination showed microtia, preauricular tag with a blind fistula anterior to right pinna. Forehead and eyes appeared symmetric but right facial length was shorter as compared to left. Cleft soft palate and bifid uvula was observed on oral examination. There was no defect of the vertebral column. Systemic examination did not show any significant abnormality. Biochemical and hematological investigations were normal. 2- Dimensional echocardiography revealed visceroatrial situs solitus, atrioventricular and ventriculoatrial concordance with 3 pulmonary veins draining into left atrium. Inferior vena cava and superior vena cava drained normally. Interatrial

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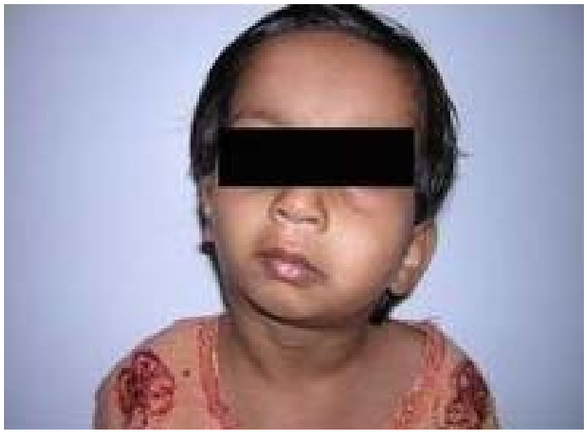

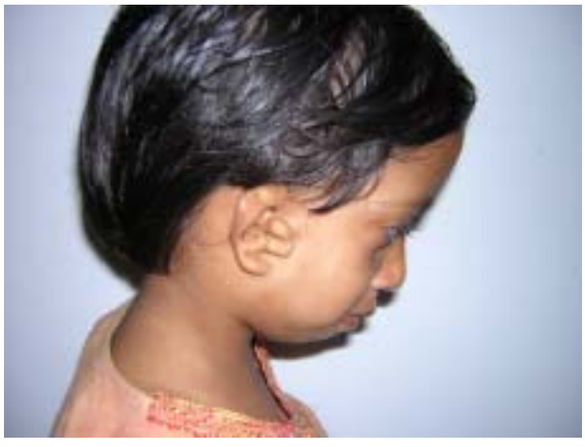

Fig 1A & 1B: Photographs of the patient showing the facial asymmetry, microtia, preauricular tag and a blind fistula.

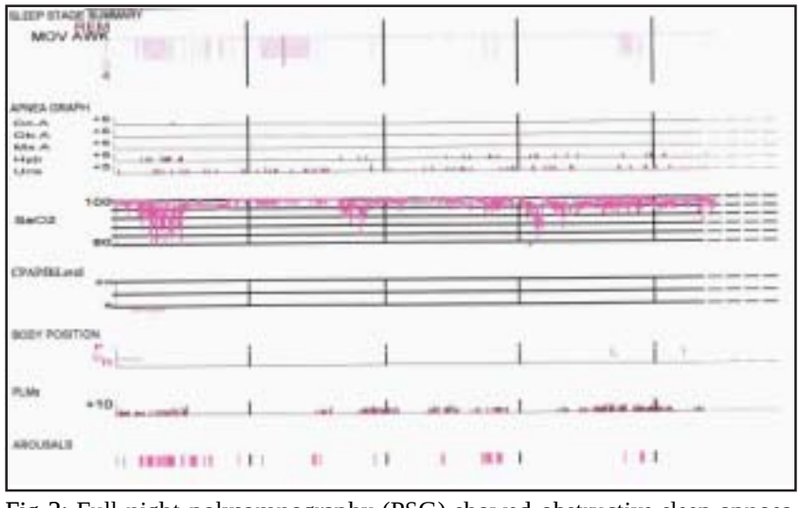

Fig 3: Full night polysomnography (PSG) showed obstructive sleep apnoea and oxygen desaturation.

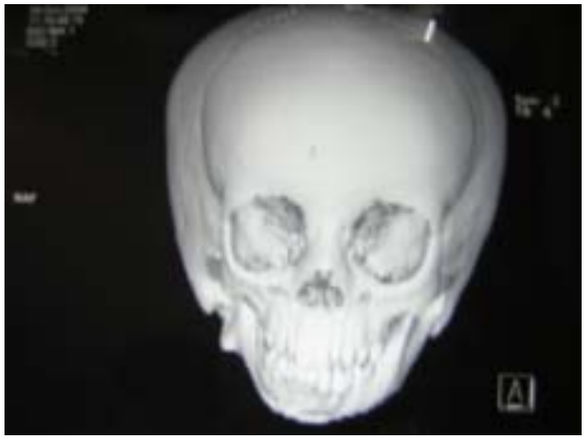

Fig 2: Three-dimensional surface reconstruction by computed tomography (CT) of face and neck showed right facial microsomia and hypoplasia of right hemimandible.

and anterventricular septae were normal. There was no evidence of pulmonary hypertension. Three-dimensional surface reconstruction by computed tomography (CT) of face and neck showed right facial microsomia and hypoplasia of right hemimandible (Figure 2), cleft soft palate and bifid uvula were also noted. Right auricle was small and inferiorly placed with atretic external auditory canal. Full night polysomnography (PSG) showed obstructive sleep apnoea was detected with respiratory disturbance index of 13.5 per hour, respiratory arousal index of 41.6 and oxygen desaturation (Figure 3).

Discussion

The most common anomalies with the Oculo-auriculo-vertebral dysplasia or Goldenhar's syndrome are hemifacial microsomia and dysplasia of the external ear affecting 65% patients.[5] Cardiovascular malformations reported include tetralogy of Fallot, coarctation of the aorta, patent ductus arteriosus, transposition of the great arteries, dextrocardia with situs solitus, atrial and ventricular septal defects, anomalous origin of the coronary arteries, right-sided descending aorta, anomalous inferior and superior venae cavae, and arrhythmic disturbances. Thirty percent of patients exhibit spine abnormalities,

varying from spina bifida, through hemivertebra, to vertebral fusion and hypoplasia.[5] Our case had anomalies of the ear (microtia), hemifacial microsomia, cardiac abnormality but no defects of the vertebral column. Recently a male infant with bilateral low-set dysplastic ears, a severe cardiac defect, rib and vertebral anomalies but without facial asymmetry has been reported.[6] The term oculoauriculo-vertebral spectrum (OAVS) is most appropriate as complex clinical presentations are seen due to variability in first and second branchial arch defects.

Our patient presented with features of OSA due to a hypoplastic mandible. Although OSA has been reported in OAVS, it is rare and is due to aplastic or hypoplastic mandible or retrognathia with an obstruction at hypopharyngeal level in children with OAVS.[4, 5] OSA in children can be diagnosed if an apnea-hypopnea index is greater than 1 or minimum oxygen saturation is less than 92 percent. This is because physiology of respiration in children differs from adults; children have higher baseline respiratory rate and apneas of three to four seconds' duration can also be accompanied by significant desaturation. Consequences of untreated obstructive sleep apnea include failure to thrive, enuresis, attention-deficit disorder, behavior problems, poor academic performance, and cardiopulmonary disease.[3] As these symptoms are nonspecific and overlap with the primary disease sleep study followed by surgical correction if necessary is an important aspect of management of OAVS. Our patient was advised surgical correction of the hypoplastic mandible at a specialized maxillo-facial surgical centre.

Author's Affiliations

Dipti Gothi, J. M. Joshi
Department of Respiratory Medicine,
T. N. Medical College and B. Y. L. Nair Hospital, Mumbai, India.

References

1. **Feingold M**, Baum J. Goldenhar's syndrome. *Am J Dis Child. 1978;132:136-8.*
2. **Tasse C**, Bohringer S, Fischer S, et al. Oculo-auriculo-vertebral spectrum (OAVS): clinical evaluation and severity scoring of 53 patients and proposal for a new classification. *Eur J Med Genet. 2005; 48:397-411. Epub 2005 Jun 8.*
3. **Chan J**, Edman JC, Koltai PJ. Obstructive Sleep Apnea in Children. *Am Fam Physician 2004;69:1147-54.*
4. **Hoch B**, Hochban W. Four-year-old girl with Goldenhar-sequence and severe obstructive sleep apnea, symptoms, diagnosis and therapy. *Int J Pediatr Otorhinolaryngol. 1998; 43:277-81.*
5. **Verona LL Nicholas**, Damian NGC, Pavarina LP, et al. Monozygotic twins discordant for Goldenhar syndrome. *J Pediatr (Rio J). 2006; 82:75-8.*
6. **McKee S**, Mabrouk R, Bali S. Bilateral microtia with severe cardiac defect: a new syndrome, or a severe manifestation of the oculoauriculovertebral spectrum? *Clin Dysmorphol. 2006;15:121-2.*

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